

# RARE DISEASE DAY

JVI

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*A joint venture with the Canadian Organization for Rare Disorders*

## 'Rare' disorders in name only

**T**hey are known as rare diseases and disorders. But they are huge in number; there may be as many as 7,750 types, according to European sources such as orphan.net. And some affect as few as 20 to 30 Canadians.

Taken as a group, however, at least one in 10 Canadians suffers from a rare disorder, and that is only the tip of the iceberg. The numbers may be much higher. Many go undiagnosed for years, so little is known of them.

"They are only rare when taken individually," says Dr. Durhane Wong-Rieger, president of the Canadian Organization for Rare Disorders (CORD). "When you consider them as a class, they have a far greater impact than common serious diseases such as diabetes, heart disease and even cancer."

And yet those who suffer these little-known disorders such as Gaucher's, Guillain-Barre, Fabry's or sickle cell anemia, often seem the forgotten ones. If they are fortunate enough to find a doctor who can diagnose their illness, they often must search to find organizations to provide them support, advice and informed counsel on how to deal with their disease.

Precious few have a ready cure. Treatment is often unavailable or so costly that provincial drug benefit plans refuse to pay for it.

"It is high time that we, as a nation, ask ourselves what values we hold," says Dr. Wong-Rieger. "I believe that, as a society, Canadians place equal value on all lives; that we believe everyone should have equal access to the treatment necessary to preserve and maintain quality of life.

"That is why we created CORD; it is the voice of all those Canadians desperately needing help facing their rare diseases and disorders."

CORD is a chorus of individual voices, a rainbow coalition for all rare disorders. It has 200 individual members, most of them members of support groups for rare diseases, and up to 30 corporate sponsors.

While it operates on a shoe-string budget, it has begun to develop considerable clout in areas such as newborn screening for genetic diseases and in advancing the cause of a made-in-Canada orphan drug policy.

"We are almost the only industrialized country that does not have in place a policy to encourage the research and development of so-called orphan drugs, ones that address rare diseases," says Dr. Wong-Rieger.

CORD is also promoting development of a national orphan drug policy. Vancouver Liberal MP Don Bell has taken up the challenge and is proposing a wide-reaching motion to



Dr. Steve Arshinoff examines Alexandra Pon. The ophthalmologist says Canada must develop a comprehensive policy on treating rare disorders.

study CORD's recommendations, with the government reporting back to Parliament on progress within six months.

Dr. Wong-Rieger notes the success of national orphan drug and rare disease policies in the United States and Europe. Canada needs to follow their lead and improve on it, she says.

The U.S. experience has shown the value of devoting time, effort and cash to rare diseases, says Stephen Groft, director of the office of rare diseases at the National Institutes of Health in Washington, D.C.

U.S. orphan drug legislation has helped bring to market 310 new products that might otherwise never have existed and has saved the lives of millions of people, he says.

"The big challenge now is getting organized to collect information on rare disease populations," he says. "With that in hand we can begin to create ways to conduct clinical studies internationally."

His office is working with 10 consortiums of researchers to help stage the studies, he says. "We are making significant headway but need a concerted effort by all stakeholders."

Canada has a rare opportunity to establish a leadership role in research and development of drugs addressing rare disorders, suggests Peter Benders, president of BIOTEC Canada, the national biotechnology industry association.

"Right now, there is an ebb and flow in many countries when it comes to drug research and development, especially where rare diseases are concerned," he says. "The United States and Europe are tightening regulations. That means if Canada adopts a comprehensive orphan drug policy, it could attract research and development here."

He points out that Canada already has 47 companies with early stage drug products in development, many of them directed at orphan diseases.

"Our problem at this moment is they are forced to take research and development offshore to countries that actively support the creation of orphan drugs," he says. "What we often forget is that there is much more than specific new therapies involved. There is the lasting intellectual capital gained through the research and development process."

The key to creating a national policy lies in raising public awareness and mobilizing support groups to actively lobby for and publicize the need, Mr. Benders says.

That is where CORD plays a vital role, says Dr. Wong-Rieger. "It is all too easy to ignore the pleas of 20 or even 1,000 people among a population of 35 million," she says. "But taken as a group, one of every 10 Canadians faces an uncertain future. Our goal is to remedy that."

### RARE DISEASE DAY

More than 20 nations, including the United Kingdom, France, Germany, Italy, Spain, Denmark, and Belgium, are celebrating the First International Rare Disease Day on Feb. 29 with royal proclamations, parliamentary debates, clinical forums, televised programs, and media events. Canada is the only official non-European participant.

Given the rarity of Feb. 29, it seemed the perfect day to raise awareness of the rare disorders and diseases and, more important, to urge international and national action to deal with these severe and often life-threatening disorders. Long neglected, these disorders are finally making their way to the forefront of medical research and clinical treatment. Some of the most innovative and exciting discoveries are happening in the areas of rare disorders but most suffer from a lack of funding and comprehensive action.

France was the first country to launch a national program on rare disorders, committing hundreds of millions of euros to the four-year initiative, which is being reviewed for renewal. The European Commission is now undertaking consultation toward a European-wide initiative.

Feb. 29 could mark the beginning of new hope for all those with rare disorders.

## CANADA FAILS RARE DISEASE VICTIMS: EXPERT

For Dr. Steve Arshinoff, rare diseases have become extraordinarily commonplace. He is an ophthalmologist with the Canadian National Institute for the Blind's Ontario Medical Mobile Eye Clinic. As such, he spends his days treating patients in remote and rural communities through Northern Ontario, communities and hamlets where no regular health care is available.

"They might never have

**'The sad fact is that we have set limits on health care budgets'**

seen an eye doctor before, or at least not in many years," he says. "The result is that I regularly see diseases and conditions that have never been diagnosed or treated, some of them quite rare."

For many, his visits can mean catching a disease in its early stages; for others, refer-

ral to a southern Ontario facility. But, for many, there is only an uncertain future. Canada's lack of a national comprehensive strategy toward diagnosis and treatment of rare diseases means there is no mechanism to pay for the expensive treatments they will need, he says.

"The sad fact is that we have set limits on health care budgets. Money goes to diseases suffered by the many, not the few," he says. "If more money is available to treat cataracts, that usually means less is available for cornea transplants."

"The money goes where the votes are."

Those diseases can run from something such as gyrate atrophy, which affects the retina and has an identified patient population of just five in Canada, to neurofibromatosis, the Elephant Man's disease, which affects a few thousand, Dr. Arshinoff says.

Elephant Man's disease causes benign tumors to spread and grow unchecked over various areas of the body, including the eyes.

See RARE on Page JV6

## A RARE DAY FOR VERY SPECIAL PEOPLE

Make Feb 29, 2008, 1st International Rare Disease Day, the start to a "Chance for Life" for Canadians with Rare Disorders.

Ask your Member of Parliament to support the Chance for Life Fund.

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Phone: (416) 969-7464 Toll Free: (877) 302-7273

  
Canadian Organization  
for Rare Disorders

www.raredisorders.ca



# RARE DISEASE DAY

*A joint venture with the Canadian Organization for Rare Disorders*

## Tragedy inspires MP

Vancouver North MP Don Bell says he has a mission. His goal: Seeing Canada create one of the world's most comprehensive orphan drug policies. The reason: He is determined to create a lasting testament to his grandson Dylan, who died last July 14 at age of 12 from primary pulmonary hypertension.

It is a rare disorder that narrows the arteries, choking off the flow of blood to the heart, brain and other vital organs. Diagnosed when he was just two, it is almost a miracle in itself that Dylan survived another 10 years.

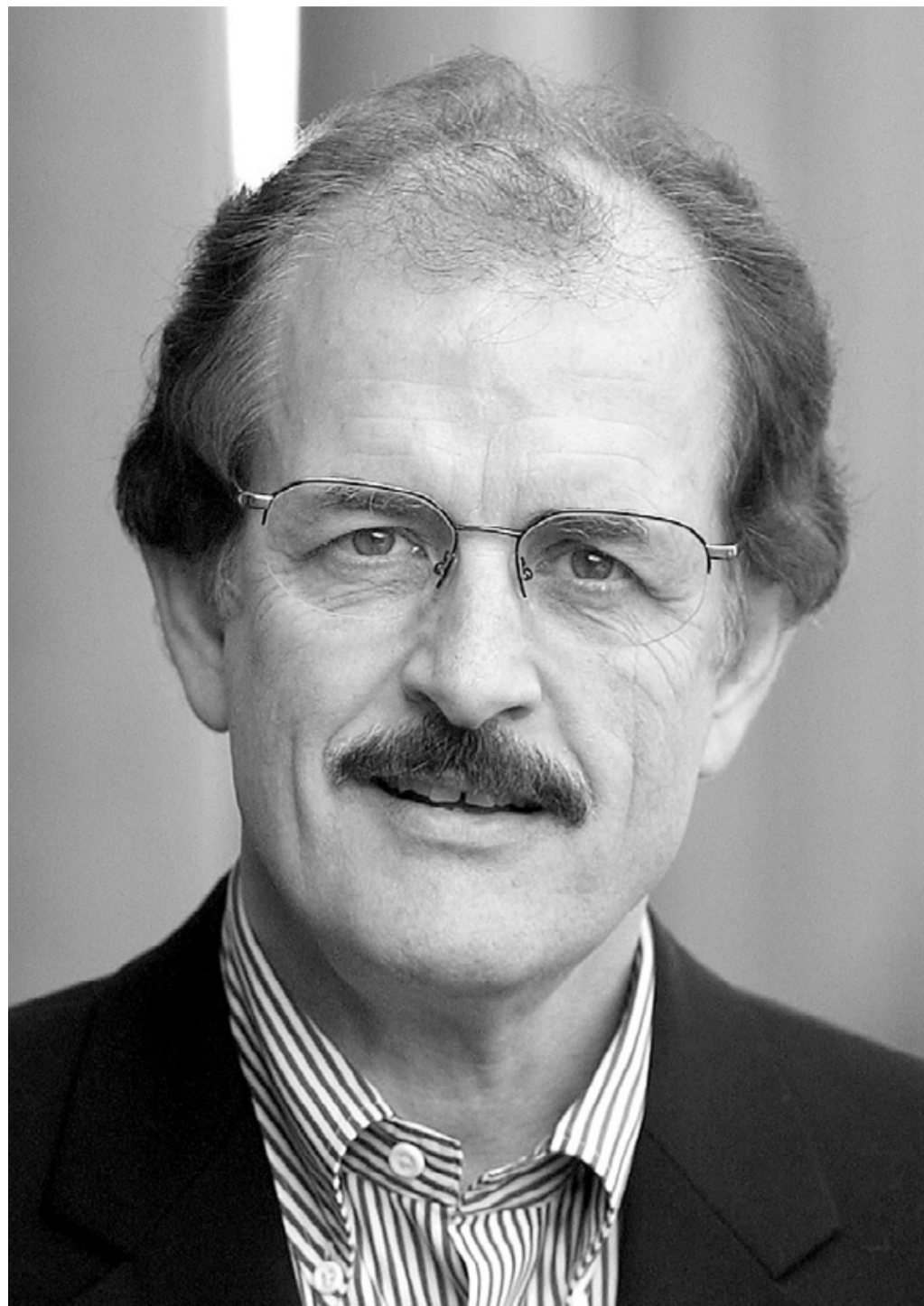
"He was a wonderful child," says Mr. Bell, 66, and a Liberal Member of Parliament for the past four years. "He was brave, funny, wonderful company. He celebrated his 12th birthday last July 13 and the day after that, collapsed in a playground.

"I want to see Canada create a policy and support structure for all those suffering rare disorders as a testament to him. It is simply something we just can no longer ignore."

The first step in what Mr. Bell concedes will likely be a long and uphill battle is a motion he introduced to the House of Commons on Dec. 6. It was to be scheduled for debate soon if his fellow MPs agree to its merit.

Carefully worded, it would commit the government to a wide-ranging study of policies and programs that would specifically address the tens of thousands of Canadians with rare disorders, of which more than 7,000 have been identified to date. The motion includes:

- Establishing an accepted formal definition of rare disorders as those affecting less than one in 2,000 Canadians.
- Examining the feasibility of the Canadian Organization for Rare Disorders' proposed national Chance for Life Fund, which would see Canada commit 2% of total annual public drug spending to rare disorders.
- Considering the creation of a multi-stakeholder advisory body to recommend treatment for patients with life-threatening or serious rare disorders.
- Considering the establishment of centres of reference for specific rare disorders to develop criteria for treatment and monitor safety and effectiveness of those treatments.
- Look at options for research and development incentives through orphan drug regulation and policy.
- Supporting international standards for conducting clinical trials in a very small patient population.
- Considering ensuring Health



MP Don Bell is lobbying to improve Canada's support system for people with rare disorders.

Canada's progressive licensing framework takes into account the differences between clinical trials for diseases affecting large patient populations and those of rare disorders, where the patient population will be significantly smaller.

■ Reporting progress on each issue back to the House of Commons within six months.

"It is a motion that both

**'One in 10 Canadians has a rare disorder. These are not isolated populations'**

sides of the House can and should support," says Mr. Bell. "It does not commit the government to spending large sums of money. Instead, it is a significant way to start the process."

Most important, perhaps, it also commits the government to report back on progress by late summer.

It was his own tragic personal experience that made Mr. Bell such a passionate advocate for treatment of rare disorders. Out of seven grandchildren, he has lost two to disease: Dylan, and his younger brother, who died two years earlier at age seven from a twisted bowel.

He has seen first-hand the great problems and hurdles

ing. He carried the intravenous kit in a little back pack that went with him everywhere. The cost was about \$20,000 a month.

"At one point we even considered a heart and lung transplant, but the doctors said all that would do was guarantee him perhaps another two years. How can a parent make decisions like that," he says.

Mr. Bell is keenly aware that the term rare disorders is misleading. Rare disorders as a class are indeed extraordinarily common.

"We have to understand that one in 10 Canadians may have a rare disorder. These are not just isolated populations," he says.

"There may, in fact, be up to 350,000 or more Canadians who desperately need this kind of policy. They need a voice and they need access to health care, just as all of us others do."

## CANADA TRAILS ON TREATMENT, RESEARCH

Despite Canada's national health care system, those suffering from rare disorders and diseases might be better off if they lived in Europe, Japan or the United States, say medical experts and patients. The reason: Access to the drugs and therapies they need to survive the diseases that ill fortune or genetics has sent their way.

Unlike most developed countries, Canada does not have a so-called orphan drug policy; a national strategy to encourage research and the development of medications designed to treat rare disorders. Canada also lacks programs to subsidize the cost of those that are already available.

"It comes down to a question of what we value as a nation," says Dr. Durhane Wong-Rieger, president of the Canadian Organization for Rare Disorders (CORD). "Costs of creating and then providing such drugs are most often cited as impediments to a national orphan drug policy.

"But we are a country that will readily pay hundreds of thousands of dollars to keep a newborn suffering a congenital condition alive and then hundreds of thousands more to treat it, even though we know the child faces years of suffering ahead. How can we then say to thousands of Canadians and their families that it just costs too much to relieve their suffering and prolong their lives?" she says.

The U.S. National Institutes of Health has identified more than 6,700 rare disorders. In some cases, such as that of sickle cell anemia, they affect thousands of people in Canada; in others, such as Fabry disease, a few hundred. The small numbers are a major hurdle preventing research into the cause, treatment and a cure.

Drug companies point out that necessary research, clinical trials and approvals processes can cost tens if not hundreds of millions of dollars. The prospects of recovering that money from such a tiny pool of patients is non-existent — without government help.

In the United States, Europe and nations such as Japan and Singapore, help has come in the form of orphan drug legislation. Governments promote research into rare disorders through a variety of grants, tax breaks and other considerations that effectively reduce the research cost burden over time.

The benefits are not only a steady stream of new medication addressing rare disorders but also the discovery that such orphan drugs can also be applied to diseases suffered by a broader base, says Dr. Marlene Haffner, executive director of regulatory affairs for Amgen Inc., the world's largest biotech company. For 20 years, Dr.

Haffner was director of the U.S. Drug Administration's office of orphan product development.

By her count, the United States has supported creation of 310 orphan drugs since the legislation was enacted in 1983, following a public outcry and demonstrations by families of those with rare diseases.

"The legislation has had an enormous beneficial impact," she says. "Those drugs now directly affect 16 million people with rare disorders and, equally important, another 48 million if you count in their families as well."

The continuing benefits of orphan drug legislation extend beyond the patients whose suffering they were designed to ease, she adds. Orphan drugs, once released, often prove effective in treating more mainstream diseases as well.

"There have been many instances where an orphan drug designed to provide relief from a specific rare disorder was then found to be useful in other diseases as well," she says. "You can't look at orphan drugs in isolation. They play an important role in so many facets of overall health care as well."

The U.S. legislation is aimed at relieving the financial burden faced by drug manufacturers through a number of incentives, which include:

- Assistance in getting initial approvals for new drugs, which has proved especially valuable in the early days of the program, Dr. Haffner says;
- Grants to university researchers to help develop proof of concept for new discoveries. Such grants are often vital to smaller companies with no revenue stream from existing products;
- Tax credits for clinical trials;
- Market exclusivity for seven years. In Europe and Japan, that exclusivity is 10 years.

"There is no dearth of rare diseases," says Dr. Haffner. "We are identifying new ones all the time. Orphan drug legislation has been of near-incalculable assistance in providing relief to patients suffering rare diseases and disorders, many of which are severe and life-threatening and affect children.

Dr. Wong-Rieger says Canada trails most of the industrialized world in orphan drug programs. She also advocates a separate federal fund similar to those available to cancer and HIV/AIDS patients in some provinces to pay or heavily subsidize the cost of orphan drugs, some of which run into the hundreds of thousands of dollars a year per patient.

"It is a matter of living up to our own beliefs," she says. "How can we tell the families of people dying from rare disorders that their life is not worth the expense of providing the drugs they need to live?"



Pompe disease is a metabolic muscle disorder first described in 1932 by Dr JC Pompe. It is a rare neuromuscular genetic disorder that occurs in babies, children, and adults who inherit a defective gene from each of their parents. The disorder has a number of synonyms, the most common are: Acid Maltase Deficiency and Glycogen Storage Disease Type II (GSD II).

The Canadian Association of Pompe was setup to help persons in Canada and elsewhere become familiar with Pompe disease, also for support of those persons with Pompe.

www.pompecanada.com  
Phone: 905-821-0623

## Canadian Neuropathy Association



### OUR MANDATE

EDUCATION, RESEARCH AND SUPPORT  
The Goals of the Canadian Neuropathy Association

To assist Neuropathy sufferers to advocate for themselves and to assist them in this process through Education and Support.

Educate the government and medical professionals to facilitate support or research into the causes and treatments of neuropathy.

To support those with peripheral neuropathy through: education, meetings, web site interaction, webinars, and by increasing the level of knowledge about this affliction.

Support investigation and research into the causes and treatment of peripheral neuropathies.

Increase public awareness about the affects of peripheral neuropathy and the need for early diagnosis and intervention.

Encourage medical insurance providers to provide appropriate coverage with regard to availability of treatments, medications and testing, as well as assistive devices.

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# RARE DISEASE DAY

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## Patients share a common bond

Adrian "Ed" Koning, 49, has Fabry's disease. He lacks an enzyme that prevents fats from accumulating unchecked in the cells of his body's organs. So far, he has suffered strokes, kidney failure, heart attacks and even a kidney transplant. The only reason he is still alive is because he receives regular enzyme replacement therapy each two weeks, at a cost of about \$250,000 a year.

Government funding for the therapy runs out in October, 2009.

Niklas Harkins, 12, of Vancouver, was diagnosed with mucopolysaccharidosis when he was five. He lacks an enzyme that rids the body's cells of noxious chemicals that build up in their walls. He is one of an estimated 300 people in Canada living with the disease, which slowly thickens tendons, joints, bones and body organs.

Niklas was one of the lucky ones, though. The British Columbia government pays the \$350,000 a year for the enzyme replacement therapy he needs. It cannot reverse damage already done but it can prevent further deterioration. Like Mr. Koning, there is no guarantee provincial funding for treatment will remain in place.

Meghan White, 18, and her sister Kate, 17, both have Gaucher's disease. It is the most common of the so-called lysosomal storage disorders. About 60 people in Canada suffer from it. Those with Gaucher's lack the enzyme that prevents fats from building up

in the body's cells and organs. Both need regular enzyme infusions to live normal lives. Again, while government programs now pay the cost, there is no program or policy in place to guarantee the future.

The funding that is available today is the result of intense lobbying and a powerful publicity campaign by the families of those with Gaucher's.

What ties these Canadians together is that they suffer from so-called rare disorders. There may be more than 7,000 disorders diagnosed to date and while some affect just scores of people globally, there are those that affect thousands. Another bond is that there is no effective drug or therapy to address most of these conditions. Patient populations are simply not large enough for drug companies to spend the tens and sometimes hundreds of millions of dollars it takes to bring new drugs to market.

The story does not end there. For those fortunate enough to have a disease where a so-called orphan drug has been developed, access may be denied because of the staggering cost of treatment. Many run into the hundreds of thousands of dollars a year.

In Canada, there is neither an orphan drug policy to encourage development of new drugs nor any reliable mechanism to ensure access for those where an effective therapy is already available. That usually means people such as Mr. Koning, Niklas Harkins and the



Ed Koning, who has Fabry's disease, joined CORD to help lobbying efforts to improve Canada's coverage of drug treatments.

White girls and their families live in a state of permanent anxiety. Their future is very uncertain, indeed.

"If I lived almost anywhere else in the world, including Brazil, the government would pay for my treatment," says Mr. Koning.

He has become active in lobbying efforts not only for those suffering from Fabry's but as vice-president of the Canadian Organization for Rare Disorders (CORD).

For Kristen Harkins, mother of Niklas and executive director of the Canadian MPS Society, Canada is as divided when it comes to who does and does not receive treatment. She notes that while B.C. children with MPS II now have access to government-funded treatment, Ontario has refused to put in place a similar program.

"It is a terrible thing that treatment depends on where you live," she says. "In effect,

provincial and federal politicians are saying your child is not worth saving."

Christine White, mother of Meghan and Kate, has also become a fierce advocate for research into and support for those suffering her children's disease. She is now president of the National Gaucher's Foundation of Canada.

"We only got financial support from the government because we did things like picket federal-provincial health

ministers' meetings and staged bedside meetings and press conferences with the minister of health," she says.

"In Ontario, if you want to receive funded treatment you must pass by a committee of physicians; they recommend who gets treatment and who does not. In Prince Edward Island, there is no funding at all. Most people can't begin to imagine what it is to know just how uncertain the future is for your children."

## CORD born from need

The Canadian Organization for Rare Disorders (CORD) is a textbook example of how dynamic organizations can grow from a good idea. Today, it is a national advocacy and education powerhouse, effectively the strong and clarion clear voice for the tens of thousands of Canadians who suffer rare disorders.

In the past, it has been all too easy for politicians, policy makers and the public at large to ignore them, passing them over in favour of focusing on diseases that affect thousands instead of scores or even hundreds. But since the support groups for hundreds of rare diseases joined forces to become a national organization in 1995, and especially since 2004, when CORD reorganized and revitalized itself, CORD has had a significant impact across all levels of policy and health care decision making.

Ask Gary Cyr, chair of the Canadian Neuropathy

Association, who joined the board in 2004, bringing with him his own group. Ask Ed Koning of the Canadian Fabry's Association, who is CORD's vice-president, or ask Durhane Wong-Rieger, CORD's president since 2005.

All are passionate about CORD's mission and its chances for future success. Simply put, they want what everyone who suffers a rare disorder wants, and what all their families want: A clear national policy that would encourage education, research, treatment and financial support for the medications vital to surviving their diseases.

"CORD grew very slowly at first," says Mr. Cyr. "In some ways, it was a wonder it survived. But thanks to the dedication of people like Maureen Gaetz-Faubert, we are a strong, vital national organization today."

It was Ms. Gaetz-Faubert who launched the predecessor

of CORD from her home in Coaldale, Alta. She suffered from a disease of the connective tissues called Ehlers-Banlos Syndrome. She quickly found that few people, including health care professionals, knew anything about it. There was nowhere to turn for information, nowhere to turn for advice on how to cope and certainly no mechanisms in place to help her deal with the symptoms and the challenges they presented to daily life.

She made a momentous decision: She would start her own support group, searching out others with Ehlers-Banlos. Together, they would form their own support network. The success of that small group grew. Others with rare disorders followed suit, forming their own support groups.

Mr. Cyr, for example, helped form the Neuropathy Association 22 years ago, also from his home. He suffered from small motor sensory

and autonomic neuropathy, which made walking difficult and caused internal organs to begin breaking down.

Ms. Gaetz-Faubert soon realized that as single groups, all these disease-specific organizations were small voices crying in a sea of indifference. She began approaching them making a persuasive case that there was strength in numbers. Hundred of voices could be a powerful choir resounding across the nation.

CORD first went province-wide in Alberta and began to attract financial support from groups such as the United Way and the provincial Lottery and Gaming Commissions.

In 1995 it became registered nationally, but it was in 2004 that its major push began.

"That is when I came on board," says Mr. Cyr. "Maureen came to me and made her case that we had to become more active, more persuasive, even have international clout. I joined at the board level."

Dr. Durhane Wong-Rieger signed on as president in 2005 and made room and staff available in her own company's

Toronto office for the national headquarters. In private life, she operates a health care consulting business. Like all of CORD's leadership, her job is performed without pay on a voluntary basis.

"Until 2006, we operated on a budget of about \$70,000 a year," she says. "Now, thanks to fundraising and growing corporate support, we are beginning to attract considerable more financing and expand our activities."

Those activities include education, lobbying, raising public awareness through the media and co-ordinating the activities of all stakeholders. CORD maintains a newsletter, it has been an effective voice in support of newborn genetic screening and has produced a white paper outlining a proposed national orphan drug policy.

That policy has been embraced by such as Liberal MP Don Bell, who has introduced a motion seeking government support in the House of Commons.

"CORD is also important because it gives us all access



Dr. Durhane Wong-Rieger

to information and materials produced by similar groups in other nations," says Mr. Cyr. "We work closely with the rare disorders groups in places like the United States and Australia. We exchange news and information on new research and new drugs."

"By sharing information, we can help all our various members understand the latest development in their own disease."

CORD's work and the importance of its place in Canadian health care is vital, says Mr. Koning. "I am not at all certain where many of us would be today without it," he says.



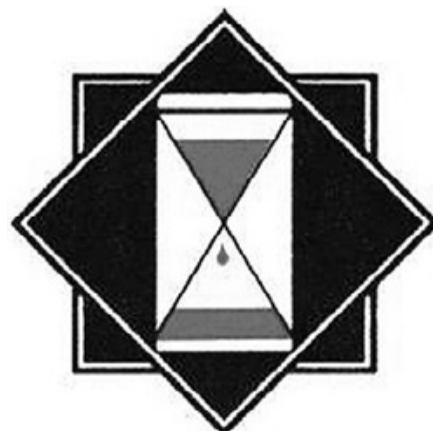
**Our Mission is to provide and promote education, research, and advocacy about anemia - causes, effects, diagnosis and treatment.**

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*A joint venture with the Canadian Organization for Rare Disorders*



Jennifer Fitzpatrick of McGill University says genetic counsellors serve a vital role in helping sufferers of rare disorders and their families cope with their afflictions.

## Front line of treatment

There are just 250 of them across Canada, and they shoulder a Herculean task. Canada's genetic counsellors face the daily challenge of informing, educating, counselling and helping diagnose rare diseases and disorders in families.

They are on the front line of treatment and often the only knowledgeable people whom families can turn to for help in understanding rare diseases and the impact on them and their loved ones.

To understand the complexity of their work and its importance, start with the number of rare diseases identified to date. The European [www.orphan.net](http://www.orphan.net) (http://www.orphan.net) site lists about 7,750, although it has information on only 3,800 of them.

Then, consider the impact their work could have on the future of families. Genetic counsellors may get involved in helping couples decide to continue with a pregnancy if the baby has tested positive for disease or defect, says Jennifer Fitzpatrick, director of the Master of Science program in genetic counselling at McGill University in Montreal. They may help find treatment, identify support groups or help patients and their

families understand what the future may hold.

Then, look toward the future. Massive gains made unravelling the mysteries of the human genome mean the medical community is beginning to understand the impact of specific chromosomes on disease. We are now able to screen at birth for hereditary conditions. We are able to do pre-natal testing for the chances a baby will carry one of those diseases. We are even able to begin screening entire

communities for susceptibility to common diseases such as type 2 diabetes.

"Genetic counselling is growing rapidly in importance, and demand is huge," says Gail Ouellette, a counsellor in private practice at Procrea Clinic in Montreal. "Rare diseases, for example, will be a growing problem for three reasons. First, we face an ageing population and some rare disorders affect the elderly more than the young.

"Second, with an increase in immigration, disorders specific

to ethnic populations, such as sickle cell anemia, are on the rise, and third is the fast pace of discovering new disorders."

One of those developments is the appearance in the United States of private laboratories, which will do genetic testing through a simple mouth swab, she points out.

"Those labs track the genome and can come back with reports that suggest things such as the patient has a 20% higher susceptibility to Alzheimers," Ms. Fitzpatrick says.

The hardest part of the job is finding support systems for patients, which can end their sense of isolation, says Ms. Ouellette. "In some cases, such as Pompe's disease, there are only 12 patients identified in Canada," she notes.

Another is finding medications and therapies that may provide relief, if not a cure.

"While there are well over

### 'Genetic counselling is growing rapidly in importance, and demand is huge'

7,000 rare disorders identified to date, programs such as the U.S. Orphan Drug program has only produced 310 drugs," she adds.

The counsellors staff 12 regional genetic centres across Canada and also maintain private practices. They work with teams of physicians, specialists, nurses and dieticians in an effort to provide one-stop solutions to patients and families with rare disorders.

A significant brake on meeting demand, however,

is that only four universities — McGill, the University of Toronto, the University of Montreal and the University of British Columbia — offer the two-year post-graduate course to train genetic counsellors, and each of them limits the class size to just four students.

"The course is so intensive and there is so much clinical work involved that none of the schools has the resources to expand the program," Ms. Fitzpatrick says.

One of the profession's current concerns is finding a way to deliver counselling to communities as well as individuals, she says. Traditionally, counselling involved 90-minute sessions with families but as the ability to screen for and identify susceptibility to more common diseases increases, those intensive sessions are proving impractical.

"We are trying pilot programs such as community screening in Chicoutimi," Ms. Fitzpatrick says. "We can now screen large numbers of people, hold information sessions for them and then do follow-up with families where susceptibility to disease has been identified."

While counsellors readily agree that Canada currently lacks an overall program to identify, diagnose, counsel and provide easy access to medication for those with rare diseases, they remain optimistic that research can, and will, make gains in the future.

"We could have a very different future for those with rare disorders made possible by science and medicine," Ms. Fitzpatrick says.

## SUPPORT GROUPS FILL IN THE GAPS

Support groups have become the mainstay for information, counselling and education for the thousands of Canadians and their families living with a rare disorder, says Dr. Durhane Wong-Rieger, president of the Canadian Organization for Rare Disorders.

With more than 7,000 rare disorders identified to date, plus newly discovered ones appearing each year, the medical profession is hard-pressed to keep up with diagnosis, treatment and providing information to patients and their loved ones. The result is that grassroot support groups spring up across Canada. As they identify others sharing their disease, those tiny groups — often started in the basement of a family home — grow, expand their services, connect with similar groups in other countries and, sometimes, become a powerful voice advocating for patients.

The growth of the Internet has made their task easier, Dr. Wong-Rieger says. So has a strong national coalition group such as CODR.

Here are some support groups making a difference: ■ **Lupus Canada** ([www.lupuscanada.org](http://www.lupuscanada.org)). Once considered rare, lupus in its varied forms now affects an estimated 50,000 people across Canada, says chief operating officer Judi Farrell. The national organization now has nine local branches and it is in the process of organizing support groups for patients and peer-to-peer training through an Ontario government grant.

"It is often called the disease with 1,000 faces," she says. "It is an auto-immune disease where the body's infection fighters turn on almost any organ. There is no known cause but women seem to be more affected than men and there are higher numbers among Aboriginals, Hispanics and Asians."

While there is no cure, physicians treat the symptoms with a variety of drugs, diet and lifestyle changes such as avoiding exposure to sunlight for some with a specific type of lupus.

■ **Canada Cystic Fibrosis Foundation** ([www.cysticfibrosis.ca](http://www.cysticfibrosis.ca)). Cystic fibrosis affects about 3,500 Canadians starting at birth, and only half live to reach their late 30s, says Cathleen Morrison, chief executive officer of the Canada Cystic Fibrosis Foundation. With this inherited disease, water does not escape from cell walls and clogs cells with chloride deposits. The body also does not produce enzymes needed to digest food.

"Patients need two hours each day for physiotherapy, exercise and medications to

address the symptoms, such as those that stimulate the pancreas, and aerosol inhalers to fight infection and clear the lungs," Ms. Morrison says. The cost can range from \$24,000 to \$60,000 a year, and not all provinces subsidize all parts of treatment.

While the Web site offers brochures and other vital information, the foundation is now looking at new ways of reaching patients such as Web casts. "The problem is that because of their susceptibility to infection, patients must avoid contact with others who have the disease or anyone who might pass an infection on to them," she says.

■ **Scleroderma Society of Canada** ([www.scleroderma.ca](http://www.scleroderma.ca)). Up to 16,000 Canadians suffer from this once relatively rare disease, says Grant Dustin, secretary-treasurer of the Scleroderma Society of Canada. "There are about as many as with multiple sclerosis," he says. Scleroderma is a hardening of the body's connective tissues and can present in almost as many ways as lupus, he says.

"There is no cure and no single treatment. You must simply deal with the symptoms," Mr. Dustin says. The society started in 1999, as local support groups began joining together to share information and resources. It now offers seven different brochures, a quarterly newsletter and annual conferences and symposiums.

■ **Guillain-Barre Syndrome Foundation of Canada Inc.** ([www.gbsfi.ca](http://www.gbsfi.ca)). Guillain-Barre sees the body's infection-fighting systems turn on the myelin sheathing that covers muscles. Unchecked, it can stop breathing or result in paralysis, says Susan Keast, a director. About 600 people in Canada have various forms, which can be a simple as a single occurrence or chronic in nature.

"The biggest need is support for patients and families," Ms. Keast says. "We now have 35 volunteers who go into hospitals, clinics and schools, and talk with patients themselves about their own experiences."

The foundation works with its international counterpart in the United States to provide brochures, DVDs and a twice-a-year newsletter. This year, the Canadian foundation is also giving a trio of \$1,500 grants to young researchers to allow them to travel to conferences of their choosing. In return, they commit to writing an article for the newsletter.

"Last year, we raised \$10,000 through a walkathon. This year, we hope to do the same, if not more, through a golf tournament. Raising funds is always crucial to groups like this," she says.

## Aplastic Anemia and Myelodysplasia Association of Canada



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**Lupus Canada is a national voluntary organization dedicated to improving the lives of people living with lupus through advocacy, education, public awareness, support and research.**

### Lupus Canada Vision:

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# The Canadian Organization for Rare Disorders

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There are up to 7,000 rare disorders affecting nearly 10% of the Canadian population. Many are serious and life threatening.

Thanks to advances in medical science, many can now be treated to improve quality of life, prolong life, and even allow some to lead near-normal lives.

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# RARE DISEASE DAY

*A joint venture with the Canadian Organization for Rare Disorders*

## 'Future up in the air'

When Dante Johnson was three weeks old, his parents noticed something was amiss. The tiny newborn had none of the vigor a baby should have. "He was floppy. That is really the only way to describe it," says his father, Keegan. "He also wouldn't eat. We knew there was something wrong; we just didn't know what."

Patricia Parker noticed similar signs with her daughter, Vicki. But that was 36 years ago. It took doctors almost three years to come up with the right diagnosis.

Both children suffer from a rare disorder called Prader Willi Syndrome. It stems from a misshapen chromosome inherited from parents. The incidence is just one in 15,000 in Canada but when it strikes, the effects can be devastating. "Most children with Prader Willi fail to grow above four feet tall and they lack muscle tone, often suffer problems with intellectual development and have almost no impulse control," says Mr. Keegan. Worst of all, they are always hungry; they have no sensation of being full. Obesity can be an inevitability.

Lifespans can also be severely shortened. While there is no cure there is treatment for some of the symptoms, such as stunted growth, through application of lifelong human growth hormones (HGH). Sadly, in Canada that treatment is not only fiercely expensive, many provincial drug plans will either not pay the cost or impose qualifying standards that are impossible for some patients to meet.

In addition, there are regular rounds of physical and occupational therapy, exercise, speech therapy and rigid dietary controls.

"My son's future is very much up in the air," says Mr. Johnson. "OHIP only pays for HGH if, when the child is tested, the hormone is lacking; that day, it was not. My wife has returned to work as a teacher and her third-party coverage now pays 80% of the cost. We don't know what will happen when he is too old and coverage no longer applies."

Currently, that cost is about \$6,000 a year. The cost of HGH treatment is based on the patient's weight. Dante is now



Patricia Parker, right, has become an active member in support programs to help people afflicted with Prader Willi Syndrome, such as her daughter, Vicki, left.

just three but as he grows, so will the cost, reaching between \$20,000 and \$30,000 a year when he is an adult.

For Ms. Parker, HGH therapy was simply not an option. When Vicki was diagnosed in the 1970s, no drugs were available to treat Prader Willi. Her child is an isolated case,

a rare instance, she says. Not only has Vicki lived to her mid-30s but through training and structured environment, she has grown to just under five feet tall and has kept her weight at about 120 pounds.

Even more amazing, Vicki lives independently in her own small mid-town apart-

ment. Her home is stocked with only the food needed for that day and she is unable to work because the temptations and stress of the workplace would likely result in uncontrolled snacking and eating.

She gets five hours a day of living support paid for by Ontario's Ministry of Community

and Social Services. "That is significant in itself," says her mother. "Most people with Prader Willi need 24-hour supervision and care."

As for HGH treatment, while it would benefit Vicki, Ontario's system of approvals for payment is a major impediment, she says.

"In the United States, it is a given that if you have Prader Willi you lack HGH," she says. "All those with the disease qualify for government subsidies. In Ontario, you must prove you lack HGH" — a fact that is obvious when one of the symptoms of the disease is stunted growth, she adds.

To help all those with Prader Willi — an estimated 2,000 in Canada — and provide support for their families, both Mr. Johnson and Ms. Parker have become deeply involved in education, research and support programs. Mr. Johnson is chairman of Prader Willi Research Canada, which, in the past two years, has raised \$250,000 to find effective treatments and potentially a cure for the disease.

Ms. Parker is on the board of the Ontario Prader Willi Association. It offers an informational Web site, brochures, a newsletter, seminars and even volunteer support staff for patients and their families.

"Our biggest concern is for the uncertainty of the future," she says. "But I have lived with that for almost 34 years. It is not something you would want any parent to face."

## Blood disorders increasing

Each four to six weeks, Maria Smith (not her real name) takes her nine-year-old daughter Krystina to a clinic for the blood transfusions she needs to live. Five nights a week, she inserts a needle into Krystina's arm, hooks her up to a pump and, for 11 hours, trickles Desferal into her body.

Desferal helps rid the body of the toxic build-up of iron that comes from the transfusions. Krystina has thalassemia, a rare inherited blood disease that affects 300 to 400 people in Ontario. Her body makes a form of red blood cells that can't carry enough oxygen.

While there is a new pill called Exjade that can replace the infusions, the cost for Krystina might be as much as \$700 a week. The Ontario government will not pay. The province's drug benefit plan says to qualify for Exjade, patients must be allergic to the less expensive Desferal. Nor will the province pay for the pump; that cost Ms. Smith \$4,000.

Krystina's tale is not uncommon. Rare blood disorders such as thalassemia, sickle cell anemia, aplastic anemia and myelodysplasia (MDS) are on the rise in Canada. An ageing population and growing immigration are major causes.

All the diseases seem to have a genetic root; some affect mainly ethnic groups and some target the elderly.

What they share, however, is a lack of funding and a lack of information to identify the condition, treat it with all the drugs available worldwide and the ability to provide support for patients and their families.

"Diseases such as MDS are no longer a very rare disorder," says Dr. Richard Wells, co-director of the MDS program at Toronto's Sunnybrook Health Sciences Centre. "It generally strikes older adults; the average age of patients is 70. We see perhaps 1,500 new cases a year and we estimate there are anywhere from 10,000 to 15,000 people across the country with the disease already, which still classifies it as a rare disorder."

"It will become a major problem in the years to come because of our ageing population yet for years, little research has been done."

Gord Sanford, 68, President of the Aplastic Anemia and Myelodysplasia Association of Canada, knows first-hand the suffering that MDS brings. He was diagnosed in 2003. His bone marrow had begun to produce deformed blood cells that could not carry enough

oxygen to the rest of the body. To further complicate the situation, his MDS caused him to get leukemia. He needs platelet and red cell transfusions twice a week to just stay alive.

"Until last month there were no drugs approved in Canada for the treatment of MDS," he says. "Even now, the treatments are often experimental and off-label. If you are young, you may find a cure by having a bone marrow transplant. For older patients, this option is usually too risky even if you could find a suitable donor. What all of us with this disease share is a terrible quality of life waiting for more research to find ways to treat this disease."

Dr. Isaac Odame sees the growing nature of treating rare blood disorders each day. He is head of the sickle cell and thalassemia program at Toronto's Hospital for Sick Children. Both diseases likely have their roots as the body's defence against malaria, he says. Why the bone marrow would start producing deformed blood cells to counter malaria is a mystery, however.

"Both spring from a recessive gene so both parents must carry that gene, but either can also act as a carrier," he says. "There is no cure, and while

there are medications to help maintain quality of life, provincial drug plans are reluctant to approve them because of cost."

There are three new drugs approved for use in the United States to treat MDS but only one has been given the go-ahead in Canada, says Dr. Wells. Revlimid, approved in January, can repair chromosomal damage in about 10% of patients eliminating the need for transfusions, he says.

"Thalidomide [Revlimid] can also work wonders and get maybe 70% of MDS patients off transfusions. The downside is that its cost is about \$10,000 a month."

To fight the structure of a health care system that tends to ignore the needs of a desperately ill few in favour of the many, patients with rare blood disorders and their families are increasingly banding together to form support groups, which educate, inform, raise funds for research and lend a hand to those with blood diseases.

"We estimate one in 15 Canadians of African descent has the sickle cell anemia gene," says Lan Timothy, president of Seed of Life Philanthropic Organization. "In countries like my native Nigeria, one in 50 have it. "There is no awareness of the disease in Canada; it is hard to diagnose and hard to get the right treatment. There is a lot of ignorance around."

## CANADA NEEDS RARE DISEASE POLICY: EXPERT

RARE

Continued from Page JV1

While the CNIB Mobile Eye Clinic cannot treat the body tumors, it can surgically remove those impairing vision.

Some diseases are silent, invisible killers while others have obvious telltale signs.

Muco polysaccharidosis, for example, thickens the bones, squares the head and produces milky white cataracts.

Corneal transplants can

**'That discovery led to a new understanding of the disease'**

cure vision problems but the root cause of the disease is still unknown and, as a result, cannot be addressed by medication.

Dr. Arshinoff is a strong supporter of the need for a rare disease policy across Canada, and not just to address the suffering of the thousands of men, women and children they affect. He

points out that the study of rare diseases can lead to a better understanding of more common ailments.

He cites the example of one patient, a woman who had lost an eye to infection as a child and sought his help for a retinoma, a benign tumor affecting vision in her remaining eye. During examination, she mentioned her son suffered from retinoblastoma, cancerous tumors on his retinas.

"I referred her to one of the leading experts on retinoblastoma in Toronto," he says. "The result was the discovery that her disease is a benign variant of the malignant form. That discovery opened the door to a whole new understanding of a disease that affects thousands."

Dr. Arshinoff is passionate about his work, keenly aware of the importance of the service CNIB provides.

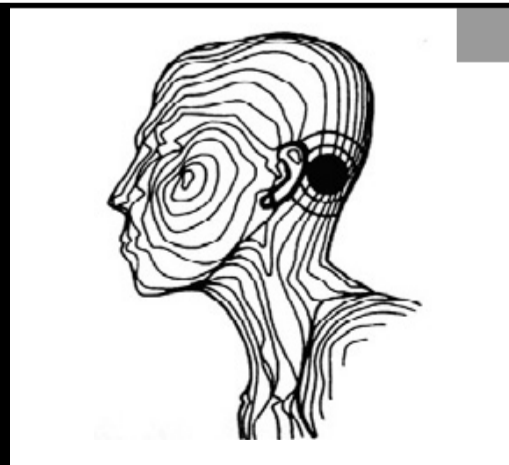
"We are always aware that if we miss something, no medical professional will likely ever see these people again. In past, if they contracted a rare disease, they often simply died. We want to prevent that from ever happening again."

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# RARE DISEASE DAY

*A joint venture with the Canadian Organization for Rare Disorders*

## CANADIAN DRUG FIRMS SLOWED BY RED TAPE

Dr. Francesco Bellini is frustrated. He is chairman and chief executive officer of Neurochem, a small Canadian pharmaceutical company. He and his team are trying to get their first major product, Eprodinate, on to the market.

At stake is not just the \$20-million invested to date but the lives of thousands of Canadians. Eprodinate has proven effective in treating people with AA Amyloidosis, a rare disease that causes proteins build up in the kidneys, clogging them and then reducing and finally stopping the kidneys' vital functions.

Those diagnosed face a five-to-15-year survival rate and in the latter stages of the disease, invariably must undergo dialysis.

The problem is that Eprodinate is a so-called orphan drug. It addresses the problems of a small sliver of the overall population, too small to attract the attention of major drug companies. While both the United States and Europe have policies encouraging development of orphan drugs, those countries have, of late, been tightening their rules.

Canada, home to a handful of young biotech and pharma companies such as Neurochem, has no orphan drug policy. As a result, Canadian companies must take advantage of orphan drug legislation abroad to develop their products and win marketing approvals.

In fact, the new drugs may never be available to Canadians. "I don't even want to talk about Canada and its position on new drugs," says Dr. Bellini. "It is just too frustrating." His current source of frustration is not just Canada but tightened demands by the U.S. Federal Drug Administration, which insists Neurochem repeat stage-three clinical tests for Eprodinate. It wants further proof of efficacy, not safety, Dr. Bellini says.

"What is lost is that patients are dying for want of this drug," he says. "We have proven it is safe, so why not get it on to the market and, if need be, retest later?" Dr. Bellini is not alone in his views. YM BioSciences Inc. of Mississauga, Ont., now has two products in clinical trials: An orphan drug called Nimotuzumab, which treats solid cancer tumors including rare tumors in the brain stems of children, and Aerolef, an inhaler that can relieve acute pain, especially post-surgical pain.

"Whether either is ever introduced to Canada is up in the air," says Sean Thompson, vice-president of corporate affairs. "There is no orphan drug policy here and no existing mechanism to pay for treatment."

Both are vital if Canadians with rare diseases and disor-

ders ever hope to get ready access to the medications and therapies they need to deal with symptoms and extend lives, Mr. Thompson and Dr. Bellini say. The cost of orphan drugs can run into the hundreds of thousands a year.

Dr. Bellini says repeating stage-three tests for Eprodinate could boost annual treatment costs from \$20,000 to \$50,000 per patient.

Smaller biotech and pharma companies are often best suited to creating orphan drugs, Mr. Thompson says. He cites his company as an example. "When we decided to form this company in 1994, we understood we could not go head-to-head with the giants such as Amgen and Bristol Myers Squibb for mass markets," he explains. "We could, however, focus on orphan drugs, an area where clinical trials are smaller and less expensive and there is support from government in the U.S. and Europe."

In YM BioSciences' case, the company licensed new discoveries — first from Cuba and then from universities around the world — and took on the task of doing development and clinical trials. Nimotuzumab is being brought to market under a licence with the Centre of Molecular Immunization in Havana.

For both companies, bringing an orphan drug successfully to market may prove just the starting point.

"The next step is to find more widespread applications," says Mr. Thompson.

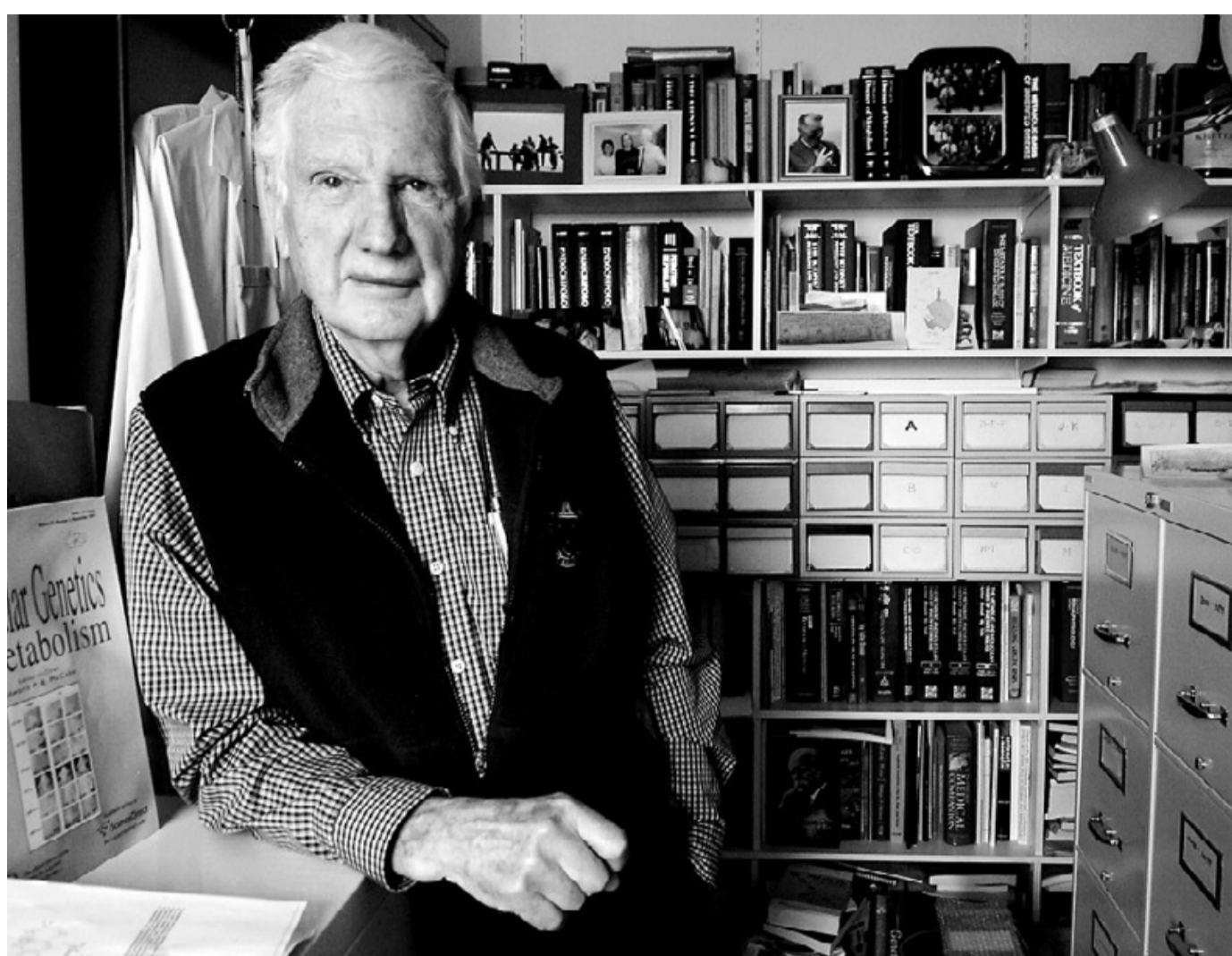
They agree that if Canada is ever to have a healthy biotech and pharmaceuticals industry, the country desperately needs an orphan drug policy that addresses both development and subsidies to patients.

"There are others like us in the same class facing the same situation," says Mr. Thompson. "The paradox is that while we are a Canadian company, we have to look to other nations to support research and development and to provide markets."

"Canadians may never see the benefits of our research."



Sean Thompson



Dr. Charles Scriver, a pioneer in the concept of newborn screening, says the process is a proven lifesaver.

## Screening for life

Six years ago, Tammy and Roger Clark's six-month old baby came down with what looked like the flu. Tammy took little Jenna to her pediatrician. Just the flu, he said. Take her home she will be all right.

At 6 a.m. the next morning, Jenna died in her mother's arms. Police in their home town of Manotick, near Ottawa, investigated the sudden death. Doctors searched for a cause. The first diagnosis was Reyes syndrome. Six weeks later, the real reason baby Jenna died was revealed. She had died of MCADD, a rare inherited disease in which the body can't oxidize fats. "We had no idea we carried that gene," says Tammy Clark. "Oh, how I wish we had."

Ms. Clark vowed that she would do her best to ensure that no other parents would face what she did. In 2004, she helped form the Save Babies Through Screening Foundation of Canada, a vocal advocate for screening newborns for a range of genetic disorders.

In 2005, for the first time in 27 years, partly through her foundation's efforts, the Ontario government announced plans to expand its newborn screening program from two diseases to 29. The 29th disease, cystic fibrosis, is being added this spring.

But the foundation still has a formidable task ahead of it,

she says. "The disparity across Canada of which provinces screen for what is enormous," she says. "Some screen for a small handful; Ontario does nearly 30. When you consider that last year alone we identified 10 babies with MCADD in Ontario, you can see how vital testing is. It saves lives."

Screening is enormously effective as a lifesaving tool, says Dr. Isaac Odame, a pediatric hematologist oncologist at Toronto's Hospital for Sick Children and head of the sickle cell and thalassemia program. "Since screening started, we have been able to reduce

ample, are prone to inherited sickle cell anemia, thought to be a natural evolution within the body to fight the effects of that mosquito born disease. Thalassemia, another blood cell condition, seems to be more prevalent in those racial and ethnic groups as well.

The screening process is simple. A small blood sample is taken from the newborn's heel and absorbed on a slip of blotting paper that is sent to a regional laboratory for testing through mass spectroscopy.

"The new technology has made mass testing possible and greatly extended the range

40,000 newborns in Quebec. Impressed by the study, then-provincial health minister Claude Castonguay provided funding for newborn genetic screening and counselling, education and treatment.

"Castonguay was one of my personal heroes," Dr. Scriver says. "We were not only able to begin identifying a wide range of diseases in newborns but also prove that some conditions we thought were diseases were not diseases at all. We were also able to offer treatment. "We saved the lives of many, many children."

Canada, however, seems to be backsliding from the days when Quebec was held up as the world leader in newborn screening and treatment.

"Unfortunately, we seem to have lost that leadership position," he says. "Not all provinces provide widespread screening and many patients can no longer be certain of funding for treatment."

Dr. Odame is another voice calling for harmonization among the provinces when it comes to screening.

"There is no need to have local laboratories; we could have regional ones to reduce cost," he says. "That is what we advocate for. Canada should take the same approach it has to diagnosing and treating cancer, an area where we have made terrific inroads."

### 'Screening will become increasingly important as the face of Canada changes'

mortality rates for children under age five for some diseases by 90%," he says. "Screening will become increasingly important as the face of Canada changes through immigration."

Where, 30 years ago, almost all immigration was from Northern Europe, new Canadians now come from Asia, Africa, the Caribbean, Central and Eastern Europe. Each of those regions has its own unique rare and not-so-rare disorders.

Those from nations where malaria is common, for ex-

ample, are prone to inherited sickle cell anemia, thought to be a natural evolution within the body to fight the effects of that mosquito born disease. Thalassemia, another blood cell condition, seems to be more prevalent in those racial and ethnic groups as well.

The screening process is simple. A small blood sample is taken from the newborn's heel and absorbed on a slip of blotting paper that is sent to a regional laboratory for testing through mass spectroscopy.

"The new technology has made mass testing possible and greatly extended the range



*The Canadian Society for  
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